

# Health Care Provider Fact Sheet

## Disease Name

## Biotinidase Deficiency

### Alternate name(s)

MULTIPLE CARBOXYLASE DEFICIENCY, LATE-ONSET  
MULTIPLE CARBOXYLASE DEFICIENCY, JUVENILE-ONSET  
BTD DEFICIENCY

### Acronym

BIOT

### Disease Classification

Metabolic Disorder

### Symptom onset

Prior to 12 months of age

### Symptoms

In the untreated state, profound biotinidase deficiency during infancy is usually characterized by neurological and cutaneous findings that include seizures, hypotonia, and rash, often accompanied by hyperventilation, laryngeal stridor, and apnea. Older children may also have alopecia, ataxia, developmental delay, neurosensory hearing loss, optic atrophy, and recurrent infections. Individuals with partial biotinidase deficiency may have hypotonia, skin rash, and hair loss, particularly during times of stress. All symptomatic children improve when treated with 5 to 10 mg of oral biotin per day.

### Natural history without treatment

Prolonged symptoms prior to institution of biotin therapy may leave the patient with varying degrees of neurological sequelae, including mental retardation, seizures, and coma. Death may result from untreated profound biotinidase deficiency.

### Natural history with treatment

If treated promptly, biotinidase deficiency may be asymptomatic.

### Treatment

Biotin supplement daily

### Emergency medical treatment

See sheet from American College of Medical Genetics (attached) or for more information, go to website:  
<http://www.acmg.net/StaticContent/ACT/Biotinidase.pdf>

### Inheritance

Autosomal recessive

### General population incidence

1:60,000 estimated with either profound or partial deficiency

### OMIM Link

<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=253260>

### Genetests Link

[www.geneclinics.org](http://www.geneclinics.org)

### Support Group

Biotinidase Family Support Group  
<http://biotinidasedeficiency.20m.com/>

Children Living with Inherited Metabolic Diseases  
<http://www.climb.org.uk/>

4-26-2010 Update